



## SPG7 gene

SPG7, paraplegin matrix AAA peptidase subunit

### Normal Function

The *SPG7* gene provides instructions for producing a protein called paraplegin, which is a member of the AAA protein family. This protein family plays a role in many cellular activities, including regulation of cell components and proteins. Located within the inner membrane of the energy-producing centers of cells (mitochondria), paraplegin is one of the proteins that form a complex called the m-AAA protease. The m-AAA protease is responsible for assembling ribosomes (cellular structures that process the cell's genetic instructions to create proteins) and removing nonfunctional proteins in the mitochondria.

### Health Conditions Related to Genetic Changes

[progressive external ophthalmoplegia](#)

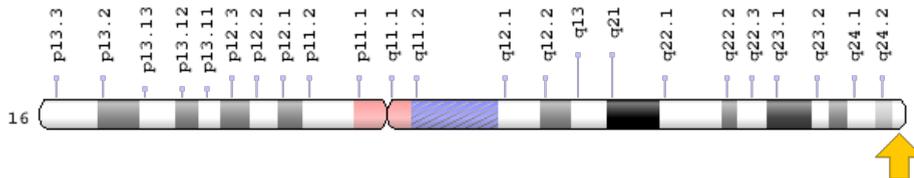
[spastic paraplegia type 7](#)

More than 10 mutations that cause spastic paraplegia type 7 have been found. Most of these mutations change single protein building blocks (amino acids) in the protein paraplegin. When paraplegin is mutated, it cannot organize with other proteins within the mitochondria to form the m-AAA protease. The buildup of unusable proteins in nerve cells, caused by the nonfunctional m-AAA protease, can impair mitochondrial functioning and diminish nerve cell signaling, leading to the major signs and symptoms of spastic paraplegia type 7.

## Chromosomal Location

Cytogenetic Location: 16q24.3, which is the long (q) arm of chromosome 16 at position 24.3

Molecular Location: base pairs 89,508,379 to 89,557,768 on chromosome 16 (Homo sapiens Annotation Release 108, GRCh38.p7) (NCBI)



Credit: Genome Decoration Page/NCBI

## Other Names for This Gene

- CAR
- cell adhesion regulator
- CMAR
- FLJ37308
- MGC126331
- MGC126332
- paraplegin, isoform 1
- PGN
- spastic paraplegia 7
- spastic paraplegia 7 (pure and complicated autosomal recessive)
- SPG5C
- SPG7\_HUMAN

## Additional Information & Resources

### GeneReviews

- Spastic Paraplegia 7  
<https://www.ncbi.nlm.nih.gov/books/NBK1107>

## Scientific Articles on PubMed

- PubMed  
<https://www.ncbi.nlm.nih.gov/pubmed?term=%28SPG7%5BTIAB%5D%29+OR+%28paraplegin%5BTIAB%5D%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+360+days%22%5Bdp%5D>

## OMIM

- SPG7 GENE  
<http://omim.org/entry/602783>

## Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology  
[http://atlasgeneticsoncology.org/Genes/GC\\_SPG7.html](http://atlasgeneticsoncology.org/Genes/GC_SPG7.html)
- ClinVar  
<https://www.ncbi.nlm.nih.gov/clinvar?term=SPG7%5Bgene%5D>
- HGNC Gene Family: AAA ATPases  
<http://www.genenames.org/cgi-bin/genefamilies/set/413>
- HGNC Gene Symbol Report  
[http://www.genenames.org/cgi-bin/gene\\_symbol\\_report?q=data/hgnc\\_data.php&hgnc\\_id=11237](http://www.genenames.org/cgi-bin/gene_symbol_report?q=data/hgnc_data.php&hgnc_id=11237)
- NCBI Gene  
<https://www.ncbi.nlm.nih.gov/gene/6687>
- UniProt  
<http://www.uniprot.org/uniprot/Q9UQ90>

## **Sources for This Summary**

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